variantyx

Genomic Unity® Constitutional Genome-Wide Copy Number Variant Analysis

CP004

Overview

The diagnostic odyssey for unexplained genetic disorders is a frustrating and costly process for patients and their families. Unnecessary delays in identifying the molecular cause of the symptoms result in potentially missed opportunities for changes in treatment for the patient as well as missed screening opportunities for family members.

Genomic Unity® Constitutional Genome-Wide Copy Number Variant Analysis takes full advantage of the Genomic Unity® single platform method, providing a full, phenotypically driven analysis of all relevant CNVs.

Method

PCR free whole genome sequencing (WGS) is used as the underlying NGS technology. Its consistent read depth across >98% of the genome enables identification of copy number variants.

Rigorously trained variant scientists interpret all variants in the context of the patient's phenotype and generate a unified clinical report.

Included analyses

✓ Genome wide copy number changes, deletions, duplications, inversions, regions of homozygosity and mobile element insertions

Test performance

Highly sensitive detection of structural variants

- 96% clinical sensitivity
- In most cases, the exact genomic coordinates (the breakpoints) of the structural variant can be determined

Accepted sample types

- Blood - optimally 5ml
- gDNA - 5µg minimum
- Saliva

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